



PTPN22 gene

protein tyrosine phosphatase, non-receptor type 22

Normal Function

The *PTPN22* gene provides instructions for making a protein that belongs to the PTP (protein tyrosine phosphatases) family. PTP proteins play a role in regulating a process called signal transduction. In signal transduction, the protein relays signals from outside the cell to the cell nucleus. These signals instruct the cell to grow and divide or to mature and take on specialized functions.

The PTPN22 protein is involved in signaling that helps control the activity of immune system cells called T cells. T cells identify foreign substances and defend the body against infection.

Health Conditions Related to Genetic Changes

autoimmune Addison disease

Graves disease

idiopathic inflammatory myopathy

juvenile idiopathic arthritis

rheumatoid arthritis

systemic lupus erythematosus

systemic scleroderma

type 1 diabetes

vitiligo

Studies have associated the R620W variation in the *PTPN22* gene with an increased risk of vitiligo, an autoimmune condition that results in patchy changes in skin coloring (pigmentation).

As with other autoimmune disorders, this variation likely affects the activity of the PTPN22 protein, making it more difficult for the body to control the immune system

and prevent it from attacking its own tissues. While the pigment loss associated with vitiligo results from the immune system attacking pigment-producing cells (melanocytes) in the skin, it is unclear what specific circumstances trigger the immune system to do so. The condition probably results from a combination of genetic and environmental factors, most of which have not been identified.

autoimmune disorders

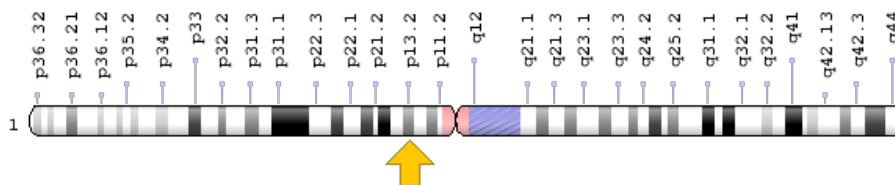
Studies have associated a variation in the *PTPN22* gene with an increased risk of several autoimmune disorders. Autoimmune disorders occur when the immune system malfunctions and attacks the body's tissues and organs. These disorders include type 1 diabetes, rheumatoid arthritis, autoimmune thyroid disease, and systemic lupus erythematosus.

The *PTPN22* gene variation associated with autoimmune disorders changes the protein building block (amino acid) arginine to the amino acid tryptophan at position 620 in the PTPN22 protein sequence, written as Arg620Trp or R620W. This variation likely affects the activity of the PTPN22 protein, making it more difficult for the body to control inflammation and prevent the immune system from attacking its own tissues.

Chromosomal Location

Cytogenetic Location: 1p13.2, which is the short (p) arm of chromosome 1 at position 13.2

Molecular Location: base pairs 113,813,811 to 113,871,761 on chromosome 1 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- hematopoietic cell protein-tyrosine phosphatase 70Z-PEP
- lymphoid phosphatase
- lymphoid-specific protein tyrosine phosphatase
- LYP
- LYP1

- LYP2
- PEP
- PEST-domain phosphatase
- protein tyrosine phosphatase, non-receptor type 8
- protein tyrosine phosphatase, non-receptor type 22 (lymphoid)
- PTN22_HUMAN
- PTPN8
- tyrosine-protein phosphatase non-receptor type 22

Additional Information & Resources

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28PTPN22%5BTI%5D%29+OR+%28protein+tyrosine+phosphatase,+non-receptor+type+22%5BTI%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+720+days%22%5Bdp%5D>

OMIM

- PROTEIN TYROSINE PHOSPHATASE, NONRECEPTOR-TYPE, 22
<http://omim.org/entry/600716>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
http://atlasgeneticsoncology.org/Genes/GC_PTPN22.html
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=PTPN22%5Bgene%5D>
- HGNC Gene Family: Protein tyrosine phosphatases, non-receptor type
<http://www.genenames.org/cgi-bin/genefamilies/set/812>
- HGNC Gene Symbol Report
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=9652
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/26191>
- UniProt
<http://www.uniprot.org/uniprot/Q9Y2R2>

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